



INTERNATIONAL APPLICATION PUBLISHED UNDER THE PATENT COOPERATION TREATY (PCT)

(51) International Patent Classification ⁶ : C12N 15/57, 9/64, A61K 38/48, G01N 33/50, C12Q 1/68, C12N 5/10, A61K 48/00	A3	(11) International Publication Number: WO 96/16175 (43) International Publication Date: 30 May 1996 (30.05.96)
(21) International Application Number: PCT/EP95/04575 (22) International Filing Date: 21 November 1995 (21.11.95) (30) Priority Data: 94402668.1 22 November 1994 (22.11.94) EP (34) Countries for which the regional or international application was filed: GB et al. (71) Applicant (for all designated States except US): ASSOCIATION FRANÇAISE CONTRE LES MYOPATHIES [FR/FR]; 13, place de Rungis, F-75013 Paris (FR). (72) Inventors; and (75) Inventors/Applicants (for US only): BECKMANN, Jacques [FR/FR]; 95, rue de Paris, F-94220 Charenton-le-Pont (FR). RICHARD, Isabelle [FR/FR]; 72, rue de l'Essonne, F-91000 Evry (FR). (74) Agents: GUTMANN, Ernest et al.; Ernest Gutmann - Yves Plasseraud S.A., 3, rue Chauveau-Lagarde, F-75008 Paris (FR).	(81) Designated States: CA, JP, US, European patent (AT, BE, CH, DE, DK, ES, FR, GB, GR, IE, IT, LU, MC, NL, PT, SE). Published <i>With international search report.</i> <i>Before the expiration of the time limit for amending the claims and to be republished in the event of the receipt of amendments.</i> (88) Date of publication of the international search report: 10 October 1996 (10.10.96)	
(54) Title: LGMD GENE CODING FOR A CALCIUM DEPENDENT PROTEASE (57) Abstract A nucleic acid sequence comprising: 1) the sequence represented in figure 8; or 2) the sequence represented in figure 2; or 3) a part of the sequence of figure 2 with the proviso that it is able to code for a protein having a calcium dependant protease activity involved in a LGMD2; or 4) a sequence derived from a sequence defined in 1), 2) or 3) by substitution, deletion or addition of one or more nucleotides with the proviso that said sequences still codes for said protease.		

FOR THE PURPOSES OF INFORMATION ONLY

Codes used to identify States party to the PCT on the front pages of pamphlets publishing international applications under the PCT.

AT	Austria	GB	United Kingdom	MR	Mauritania
AU	Australia	GE	Georgia	MW	Malawi
BB	Barbados	GN	Guinea	NE	Niger
BE	Belgium	GR	Greece	NL	Netherlands
BF	Burkina Faso	HU	Hungary	NO	Norway
BG	Bulgaria	IE	Ireland	NZ	New Zealand
BJ	Benin	IT	Italy	PL	Poland
BR	Brazil	JP	Japan	PT	Portugal
BY	Belarus	KE	Kenya	RO	Romania
CA	Canada	KG	Kyrgyzstan	RU	Russian Federation
CF	Central African Republic	KP	Democratic People's Republic of Korea	SD	Sudan
CG	Congo	KR	Republic of Korea	SE	Sweden
CH	Switzerland	KZ	Kazakhstan	SI	Slovenia
CI	Côte d'Ivoire	LI	Liechtenstein	SK	Slovakia
CM	Cameroon	LK	Sri Lanka	SN	Senegal
CN	China	LU	Luxembourg	TD	Chad
CS	Czechoslovakia	LV	Latvia	TG	Togo
CZ	Czech Republic	MC	Monaco	TJ	Tajikistan
DE	Germany	MD	Republic of Moldova	TT	Trinidad and Tobago
DK	Denmark	MG	Madagascar	UA	Ukraine
ES	Spain	ML	Mali	US	United States of America
FI	Finland	MN	Mongolia	UZ	Uzbekistan
FR	France			VN	Viet Nam
GA	Gabon				

INTERNATIONAL SEARCH REPORT

International Application No
PCT/EP 95/04575

A. CLASSIFICATION OF SUBJECT MATTER

IPC 6 C12N15/57 C12N9/64 A61K38/48 G01N33/50 C12Q1/68
C12N5/10 A61K48/00

According to International Patent Classification (IPC) or to both national classification and IPC

B. FIELDS SEARCHED

Minimum documentation searched (classification system followed by classification symbols)

IPC 6 C12N A61K G01N C12Q

Documentation searched other than minimum documentation to the extent that such documents are included in the fields searched

Electronic data base consulted during the international search (name of data base and, where practical, search terms used)

C. DOCUMENTS CONSIDERED TO BE RELEVANT

Category *	Citation of document, with indication, where appropriate, of the relevant passages	Relevant to claim No.
P,X	CELL, APR 7 1995, 81 (1) P27-40, UNITED STATES, XP002010548 RICHARD I ET AL: "Mutations in the proteolytic enzyme calpain 3 cause limb-girdle muscular dystrophy type 2A." see the whole document	1-20
X	--- JOURNAL OF BIOLOGICAL CHEMISTRY, vol. 264, no. 33, 1989, BALTIMORE, MD US, pages 20106-20111, XP002010549 HIROYUKI SORIMACHI ET AL.: "Molecular cloning of a novel mammalian calcium-dependent protease distinct form both m- and mu-types" cited in the application see figures 2,3 --- -/--	1-8, 12-15

☒ Further documents are listed in the continuation of box C.

☐ Patent family members are listed in annex.

* Special categories of cited documents:

- *A* document defining the general state of the art which is not considered to be of particular relevance
- *E* earlier document but published on or after the international filing date
- *L* document which may throw doubts on priority claim(s) or which is cited to establish the publication date of another citation or other special reason (as specified)
- *O* document referring to an oral disclosure, use, exhibition or other means
- *P* document published prior to the international filing date but later than the priority date claimed

T later document published after the international filing date or priority date and not in conflict with the application but cited to understand the principle or theory underlying the invention

X document of particular relevance; the claimed invention cannot be considered novel or cannot be considered to involve an inventive step when the document is taken alone

Y document of particular relevance; the claimed invention cannot be considered to involve an inventive step when the document is combined with one or more other such documents, such combination being obvious to a person skilled in the art.

& document member of the same patent family

Date of the actual completion of the international search

14 August 1996

Date of mailing of the international search report

20.08.96

Name and mailing address of the ISA

European Patent Office, P.B. 5818 Patentlaan 2
NL - 2280 HV Rijswijk
Tel. (+31-70) 340-2040, Tx. 31 651 epo nl,
Fax (+31-70) 340-3016

Authorized officer

Espen, J

INTERNATIONAL SEARCH REPORT

International Application No

PCT/EP 95/04575

C.(Continuation) DOCUMENTS CONSIDERED TO BE RELEVANT

Category *	Citation of document, with indication, where appropriate, of the relevant passages	Relevant to claim No.
X	JOURNAL OF BIOLOGICAL CHEMISTRY, vol. 268, no. 26, 1993, BALTIMORE, MD US, pages 19476-19482, XP002010550 HIROYUKI SORIMACHI ET AL.: "A novel tissue-specific calpain species expressed predominantly in the stomach comprises two alternative splicing products with and without Ca ²⁺ -binding domain" see figure 2 ---	1-8, 12-15
X	BIOCHEMISTRY, vol. 27, 1988, EASTON, PA US, pages 8122-8128, XP002010551 SHINOBU IMAJOH ET AL.: "Molecular cloning of the cDNA for the large subunit of the High-Ca ²⁺ -Requiring form of human Ca ²⁺ -activated neutral protease" cited in the application see figure 2 ---	1-8, 12-15
Y	HUMAN MOLECULAR GENETICS, vol. 3, no. 2, February 1994, pages 285-293, XP002010552 FOUGEROUSSE F. ET AL.: "Mapping of a chromosome 15 region involved in limb girdle muscular dystrophy" cited in the application see the whole document ---	1-20
Y	JOURNAL OF MEDICAL GENETICS, vol. 30, 1993, pages 385-387, XP000578358 PASSOS-BUENO M. R. ET AL.: "Evidence of genetic heterogeneity in the autosomal recessive adult forms of limb-girdle muscular dystrophy following linkage analysis with 15q probes in Brazilian families" see the whole document ---	1-20
Y	GENOMICS, OCT 1994, 23 (3) P619-27, UNITED STATES, XP000578720 RICHARD I ET AL: "Regional localization of human chromosome 15 loci." see page 625 ---	1-20
A	INT. J. BIOCHEM., vol. 22, no. 8, 1990, pages 811-822, XP002010554 JOHNSON P. ET AL.: "Calpains (intracellular calcium-activated cysteine proteinases): structure-activity relationships and involvement in normal and abnormal cellular metabolism" see the whole document ---	
	-/--	

INTERNATIONAL SEARCH REPORT

International Application No
PCT/EP 95/04575

C.(Continuation) DOCUMENTS CONSIDERED TO BE RELEVANT

Category *	Citation of document, with indication, where appropriate, of the relevant passages	Relevant to claim No.
A	<p>FASEB JOURNAL, vol. 8, no. 11, 1994, pages 814-822, XP002010555 TAKAOMI C. SAIDO ET AL.: "Calpain: new perspectives in molecular diversity and physiological-pathological involvement" see the whole document -----</p>	



- - -



2